Introduction
Persistent hyperplastic primary vitreous (PHPV), also known as persistent fetal vasculature (PFV), is a rare developmental disorder of the eye due to incomplete regression of the embryonic hyaloid vasculature. PHPV occurs due to a mutation in the ATOH7 gene on chromosome 10q21.3 with autosomal recessive and autosomal dominant inheritance described. PHPV is associated with ocular, intracranial, and neurological abnormalities, and several rare genetic syndromes (Table 1).

Case report
35 y old G3P1 presented for a scheduled third trimester scan at 34w2d with normal earlier screening and imaging. A triangular echogenic mass was identified behind the lens of the eye (fig. 1A,B). The lens itself was sonolucent, fetal eye movements were observed, and there was no increased vascularity. Fetal anatomy and growth was normal. A provisional diagnosis of PHPV was made with findings supported by fetal MRI (fig. 1C).

Postnatal findings
Following paediatric ophthalmology review, a diagnosis of posterior PHPV and retinal dysplasia was made (fig. 2) with a poor prognosis for vision. Molecular genetic testing of vitreoretinopathies was negative. Surgery was not performed and a scleral shell was fitted.

Figure 1. Axial prenatal ultrasound images through fetal orbits demonstrating an echogenic mass in the right eye with features consistent with a PHPV. Image A, demonstrates the triangular shape of the PHPV with the apex extending posteriorly to the optic disc (arrow) and video clip B, demonstrates a normal sonoluent lens. Image C, Axial T2 HASTE through fetal orbits. The lens demonstrates a low signal intensity and the globe a high signal intensity. The PHPV is identified as a triangular mass posterior to the lens with a low signal intensity (arrow). PHPV, persistent hyperplastic primary vitreous; R, right.

Figure 2. Postnatal ultrasound of the right eye, a diagnosis of PHPV was made with findings supported by retinal dysplasia. Sonographic features consistent with a PHPV are shown by Doppler imaging.

Discussion
PHPV typically presents in the neonate or infant with variable clinical features including leukokoria, cataract, microphthalmia, vitreous haemorrhage, secondary glaucoma, and congenital retinal detachment. PHPV may be classified as anterior, posterior and combined forms, according to the affected intraocular structure. Diagnosis can be difficult due to the variable clinical presentation. High frequency ultrasound, MRI, and CT have a role in establishing differential diagnosis, particularly in excluding retinoblastoma. Untreated PHPV may result in ocular haemorrhage and secondary glaucoma. Published reports of prenatal diagnosis of PHPV are typically in association with sonographically detectable intracranial abnormalities.

This case is an unique presentation of isolated unilateral PHPV. Prenatal diagnosis enabled appropriate counselling and timely postnatal management.

References